Opis choroby *

Definicja

A rare form of mucopolysaccharidosis characterized by abnormal storage of hyaluronan in lysosomes due to deficiency of hyaluronidase 1. Clinical manifestations include knee and/or hip pain associated with swelling, diffuse joint involvement with proliferative synovitis and occurrence of multiple periarticular soft-tissue masses, short stature, and dysmorphic craniofacial features (such as flattened nasal bridge, bifid uvula, and cleft palate).

Dane

Klasyfikacja
Choroba

Synonimy
MPS9
MPS9
MPSIX
Mukopolisacharydoza typu 9
Mukopolisacharydoza typu IX
MPSIX
Mucopolysaccharidosis type 9
Mucopolysaccharidosis type IX

Kod ORPHA
67041

Kod OMIM
601492

Kod ICD10
E76.2

Kod ICD11
5C56.3Y

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*Źródło

orphanet